

Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)

A fatty acid oxidation disorder

What is it?

Short Chain Acyl-CoA Dehydrogenase Deficiency (also known as SCAD) is an inherited fatty acid oxidation disorder. Patients with fatty acid oxidation disorders, like SCAD, cannot properly breakdown fats to energy. Once the body uses up its primary source of energy (glucose, or blood sugar), the body begins to fail because it cannot then make energy from fats. Therefore, people with SCAD must eat on a very regular basis and should not go long without food.

What are the symptoms?

The symptoms of SCAD can be very variable. People with SCAD can appear normal at birth. It is also possible to have episodes of low blood sugar, poor appetite, vomiting, lack of energy, shortness of breath, or seizures. Some people with SCAD may show developmental delay and low muscle tone. Many symptoms of SCAD can be prevented by immediate treatment and lifelong management. People with SCAD typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

SCAD is inherited in an autosomal recessive manner. This means that for a person to be affected with SCAD, he or she must have inherited two non-working copies of the gene responsible for causing SCAD. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have SCAD. Typically, there is no family history of SCAD in an affected person. SCAD is a rare fatty acid oxidation disorder; the total number if people affected with SCAD is not known.

How is it detected?

SCAD can be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

SCAD is treated by avoiding fasting and eating frequently, and sometimes special medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a

genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://ghr.nlm.nih.gov/ghr/page/Home>

Save Babies Through Screening Foundation

4 Manor View Circle Malvern, PA 19355-1622 Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545 Email: email@savebabies.org

Website: <http://www.savebabies.org/diseasedescriptions.php>

FOD (Fatty Oxidation Disorder) Family Support Group

1559 New Garden Rd, 2E Greensboro, NC 27410 Phone: (336) 547-8682 [8am - 8pm EST every day] Fax: (336) 292-0536 [email/call ahead between 8am and 8pm before faxing] Email: deb@fodsupport.org Website: <http://www.fodsupport.org>

United Mitochondrial Disease Foundation

8085 Saltsburg Road, Suite 201 Pittsburgh, PA 15239 Phone: (412) 793-8077 FAX:

(412) 793-6477 email: info@umdf.org website: <http://www.umdf.org/>

STAR-G Hawaii Department of Health

<http://www.newbornscreening.info/Parents/fattyaciddisorders/SCADD.html>